Suggested Follow-up for Classical Galactosemia
Deficient GALT with Normal Total Galactose

Possible Causes: Classical galactosemia is a condition of abnormal galactose metabolism caused by deficient functioning of galctose-1-P-uridyl transferase (GALT). When GALT is moderately deficient, the infant may have Duarte galactosemia or maybe a carrier for a form of galactosemia. GALT can also be falsely deficient if specimen is exposed to extremes of heat and/or humidity.

Next Steps if Abnormal: See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. If the mother is breastfeeding, it is not necessary to switch to soy based infant formula since the total galactose is within normal limits. Repeat GALT and total galactose screening on filter paper and send to the DHEC laboratory. If GALT is still deficient in repeat specimen, further testing is necessary to clarify the diagnosis.

Neonatal Presentation: Classical galactosemiað hypoglycemia, jaundice, sepsis, failure to thrive. Duarte galactosemiað none.

Emergency Treatment: Usually none.

Standard Treatment: Classical galactosemiað galactose restricted diet for life. Duarte galactosemiað usually none past infancy.

Advice for Family: Provide basic information about classical galactosemia. The handout, *When Baby Needs a Second Test for Classical Galactosemia*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html

http://www.geneclinics.org/profiles/galactosemia

http://www.genetests.org/query?dz=galactosemia

http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm